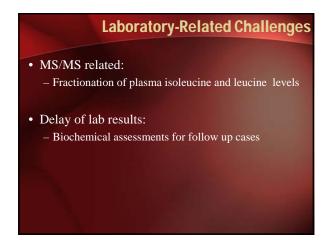




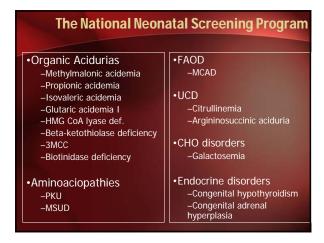
### Professionals-Related Challenges Inadequate assessment of neuropsychological outcome Inadequate professional education/awareness: Delay in instituting treatment Generalization of the false impression of poor outcome in IMD: Delay of prompt and properly aggressive ICU interventions

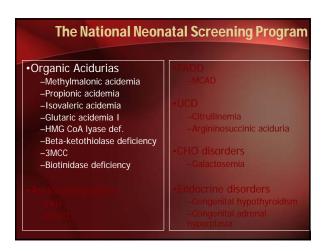


### Shortage/Unavailability of biochemical diagnostic facilities Confirmatory tests are not available Enzyme assay Metabolites (pterins) Complementation studies (MMA) Mutation detection









### • Confirmatory tests are not available - Enzyme assay - Complementation studies (MMA) - Mutation detection • B<sub>12</sub> responsiveness assays (MMA) - in-vivo and in-vitro

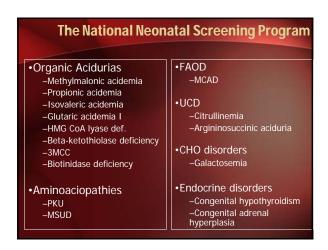
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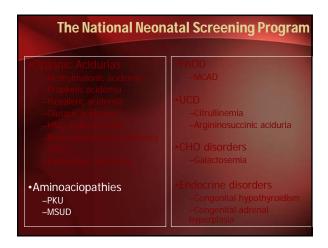
### • Carnitine / Biotin / Glycine: - Availability - Physicians knowledge (ER physicians, Pediatricians) • Bicarbonate: - improper correction of severe metabolic acidosis: • cerebral edema • prolongation of imbalanced acid-base status



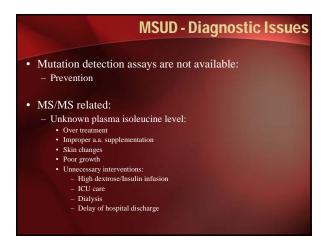
### Organic Acidurias - Complications Chronic: Neuropsychological outcome: Shortage of pediatric psychiatrists and psychologists to assess neuropsychological outcome Inadequate neurological evaluations for CNS deficits Lack of psychosocial support and interventions Renal complications: Lack of interest/experience of adult nephrologists to follow organic acidurais cases. The burden of kidney transplantation: The Priority



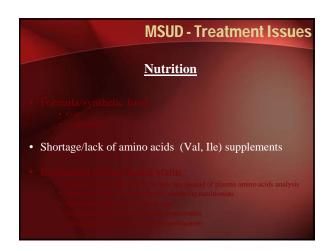




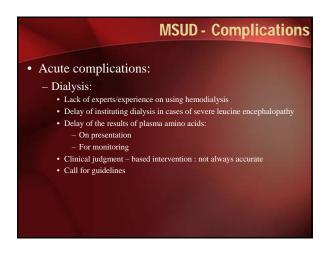




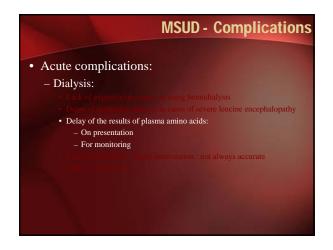




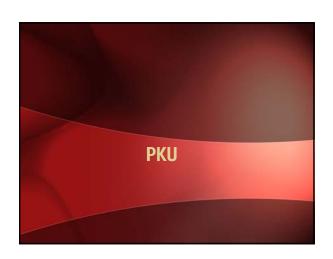




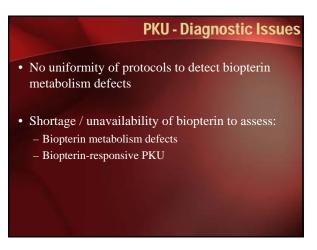


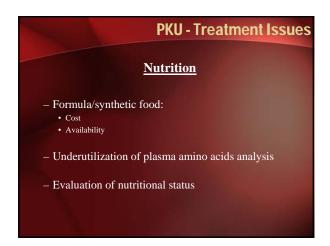




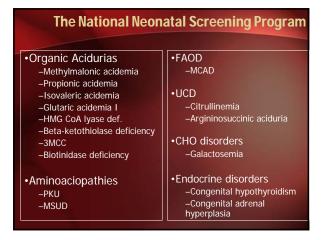


### • Enzyme / Pterine measurements / Mutation detection assays are not available: - Confirmation of the hyperphenylalaninemia variant - Prevention









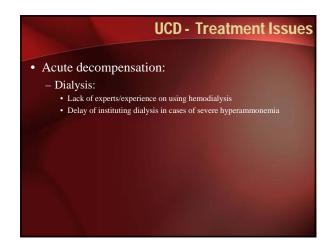


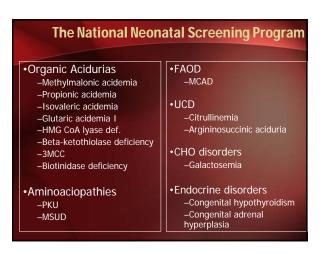
### UCD - Diagnostic Issues Enzyme and mutation detection assays are not available: Confirmation Prevention Spurious serum ammonia result – Unnecessary/Delay of interventions: ICU care Dialysis Delay of hospital discharge







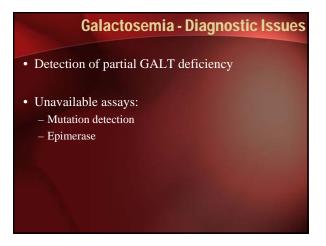














## Family/Social - Related Challenges Inadequate parents/family education Compliance issues Implication of a "Genetic" disease on the family/tribe Inadequacy of counseling services No support groups

### Family/Social - Related Challenges Inadequate services for other preventive measures: Prenatal diagnosis Preimplantation genetic diagnosis



### **System-Related Challenges**

- Maintaining the chain is a major undertaking:
  - Time of sampling/early discharge
  - Tracking of positive cases
  - Follow up of detected cases
- Efficiency of communication faces hurdles
- The wide geographic area poses a great challenge to every single element of the Program

### **System-Related Challenges**

- Inadequate evaluation of nutritional status:
  - Shortage of specialized/dedicated metabolic nutritionists
  - Unavailability of dietary products (semisynthetic diet, mixtures of a.a., modified protein hydrolysates)
  - Inadequate frequency of follow ups
  - Inadequate family/nutritionist communication
  - Inadequate nutritional laboratory assessments

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### **System-Related Challenges**

- Inadequate assessment of neuropsychological outcome:
  - Shortage of pediatric psychiatrists and psychologists to assess neuropsychological outcome
  - Inadequate neurological evaluations for CNS deficits
  - Lack of psychosocial support and interventions
- No plan for multidisciplinary approach

### • Lack of: - Treatment guidelines - Consistency of management interventions - Consensus on outcome measures

### • The infrastructure of the health care system and integrating the Screening Program





### Vision of the Program administration • The comprehensiveness of the Program —Integration of follow-ups into the Program • Pace of the Program expansion: • New disorders • New centers • New labs • ? who evaluates/plans

# Administration-Related Challenges • Funding issues: - Screening (new technologies - Diagnosis - Management (formulas / medications) - Follow up and evaluation - Education • Poor involvement of the public/media • Bureaucracy – not unexpected!



### Despite these challenges: A great opportunity still remains to overcome these issues The Program is still in its first stages. The high ambition, perseverance, and determination. Having the political support and decision has always been attainable.

